DNA motifs for identifying, isolating and assembling the nucleotide sequence of a chromosome centromere

KEYWORDS

- ❑ CENTROMERE
- ❑ HUMAN DNA
- ❑ GENOME ASSEMBLY
- ❑ GENETIC BIOMARKER
- ❑ FINGER PRINTING

AREA

- ❑ BIOMEDICAL

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Patent for invention.

Ownership / Co-Ownership
Sapienza University of Rome.

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Industrial & Commercial Reference
Biotechnologies, Genetic start-up, Computational genome assembly technologies.

Time to Market
TRL is 5.

Availability
Research, Develop-ment, Experimentation, Collaboration, Start-up and Spin-off.

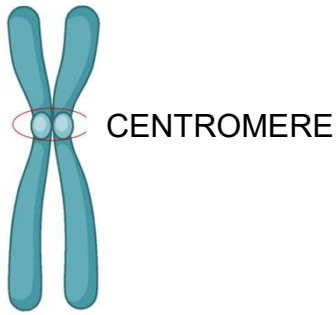

Fig 1. Human chromosome and centromere site

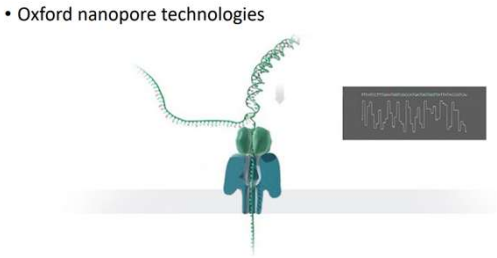

Fig 2. Nanopore long reads sequencing

Abstract
The invention concerns DNA motifs for identifying, isolating and assembling the nucleotide sequence of a chromosome centromere, in particular, for assessing the nucleotide sequence of a chromosome's centromere and the entire human genome. The invention encompasses methods for validation and analyses of these sequences in physiological and pathological states. This innovation stands to make significant contributions to the fields of genomic research and diagnostic genetics, offering a novel approach to centromere study and genome assembly.

Technical Description

The present invention represents a novel method and associated computational tools designed for analyzing human chromosomes and centromeres. The use of the patented DNA motifs used as described by the inventors represents marker for characterizing, identifying, isolating, assembling or validating the nucleotide sequence of the centromere of a human chromosome.

Technologies & Advantages

Complete genome reconstruction is technically challenging, also in terms of costs and resources. Our method allows you to assemble, validate and analyze centromeres, among the most complex regions of the human genome. The method is applicable to thousands of samples and is based on the use of sequencing technologies that were not available before 2015 but are starting to become more common in biomedical research and clinics. In the short term, this method will lead to new algorithms and software tools that can be used routinely for biomedical research, especially with a focus on human diseases associated with centromeres. Once validated in large cohorts, application for diagnostic purposes by genetic analysis laboratories (e.g. for cancer research) will follow, with the market for genetic testing constantly growing.

Applications

The method has several applications:

-Genetic and Genomic Codification of Human Centromeres and whole genome.
-Extraction of Long DNA Sequencing Reads from Centromeres
-Chromosomal Origin Determination of DNA Reads
-Validation of Centromere Sequences and Human Genome Assemblies
-Guidance for Correct Assembly of Human Centromeres and Genomes
-Identification of Aberrations and Structural Variations in Centromere DNA,
-Prediction of Active Centromere Size.
-Rapid Identification of Genome Assembly Errors

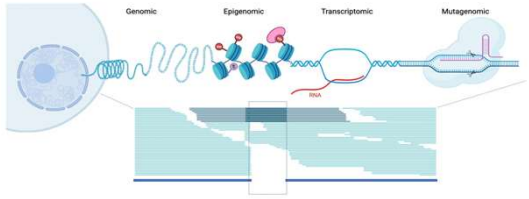

Fig. 3. Assembling reads

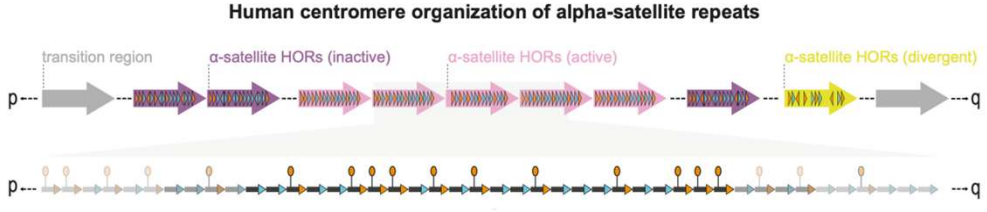

Fig.4 Method

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